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BART'S SYNDROME WITH EAR MALFORMATION AND NATAL TEETH: A CASE REPORT

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ABSTRACT

Bart's syndrome is characterized by congenital localized absence of skin, congenital epidermolysis bullosa, and associated with nail abnormalities. It is an autosomal dominant disorder with variable presentation. We report a very rare case of Bart's syndrome with multiple abnormalities. A male late preterm born to consanguineous couple, with history of similar problem in the elder sibling which died in neonatal period our case presented with absence of skin in scalp, face, trunk and extremities. Baby has deformed fingernails and eyes; ears and nose are not well formed. Peg shaped deformity of teeth, left multi cystic dysplastic kidney were noted. Baby died on the first day of life. The diagnosis of Bart's syndrome was made on clinical presentation, family history and skin biopsy.

KEYWORDS: Bart's Syndrome, Malformed Ears, Peg Shaped Natal Teeth

INTRODUCTION

Bart syndrome is one type of autosomal dominant disorder characterized by congenital localized absence of skin associated with Epidermolysis Bullosa (EB) [1]. Bart's syndrome has variable clinical manifestations [2]**.** Dystrophic epidermolysis bullosa and aplasia cutis congenita, also known as congenital localized absence of skin are rare clinical entities. Aplasia cutis congenita presented in conjunction with simplex, junctional, or dystrophic types of epidermolysis bullosa is classified as type 6 ACC (aplasia cutis congenita) [3]**.** This association was initially described and referred to in the literature as Bart Syndrome. Diagnosis of Bart's syndrome is based on cutaneous, ear and nail abnormalities [4]. Ours is a district level government teaching hospital. We report a case of Bart's syndrome, an exceedingly rare disorder with different clinical manifestations. Very few cases are reported till now in the literature.

CASE REPORT

A male baby was born to a 22 year old second gravid mother with 36 weeks of gestation. He was the second child of the consanguineous couple. Both parents are apparently healthy and had no abnormalities of skin and mucous membrane. There was positive family history of first female baby died due to similar problems within few days of birth. Mother had regular antenatal check up and did not have any history of medical problems during pregnancy. The baby was

diagnosed ante-natally of left multi cystic dysplastic kidney. Triple test was normal. Baby was born by cesarean section due to decreased fetal movements. APGAR score was 3 at 1 minute and 5 at 5 minutes. Birth weight was 1.9 kg (Small for Gestation), length was 44 cm and head circumference was 30cm. He was resuscitated with bag and mask ventilation, but eventually the baby died at 2 hours of life. On examination the baby had absence of skin over scalp, face, neck, trunk, genital region and over upper and lower extremities, Figure 1 and 2. Bilateral corneal haziness, peg shaped teeth were noted and ears are not well formed, Figure 1. The fingernails were dystrophic and deformed. There was a palpable mass at left lumbar region. Microscopic skin biopsy report suggestive as abrupt absence of epidermis, dermal portion with adnexal glands, features favoring congenital absence of skin, a variant of epidermolysis bullosa (Bart's syndrome)

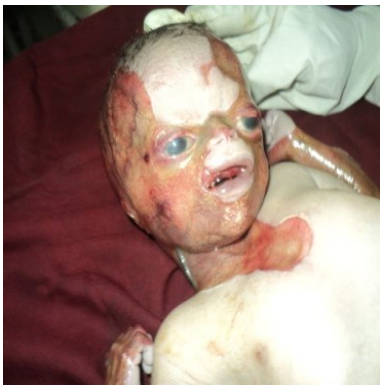

Figure 1: Natal Peg Shaped Teeth and Corneal Haziness

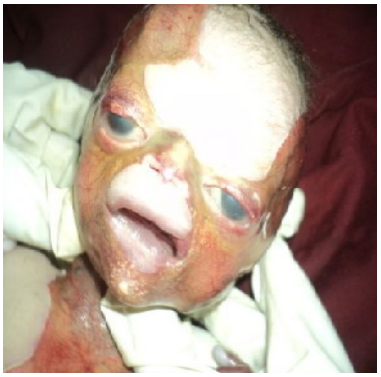

Figure 2: Deformed Ears

Figure 3: Absence of Skin over Facial Area and Hands

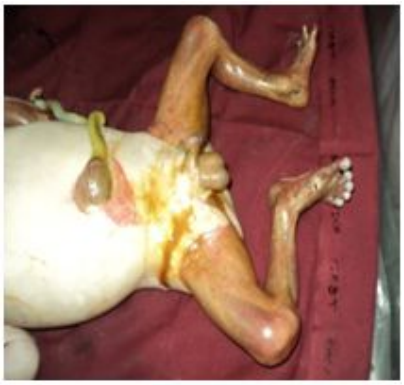

Figure 4: Absence of Skin over Lower Limbs, Umbilical Area

DISCUSSIONS

Bart's Syndrome was first described in 1966 when Bart reported a family of twenty-six members who were born with congenital absence of skin on lower extremities and blistering of skin, mucus membrane and nail dystrophy was noted in all cases. This unique association came to be known after his name as Bart's syndrome. Complete penetrance was noted in all the cases [5]. Bart considered the congenital absence of skin as an occasional manifestation of Epidermolysis bullosa simplex and attributed it to in utero blistering [6]**.** Another family described by Kanzler [7] showed the same clinical picture in four generation & was diagnosed to have epidermolysis bullosa simplex with congenital localized absence of skin. Our case presented with the same skin manifestation as that described in both families with elder sibling having same problem and died after birth. Our diagnosis was based on the clinical picture and skin biopsy. Mc Kinster suggested that congenital absence of skin in Bart's syndrome may follow the lines of blaschko [8]**.** They reported six cases in which they observed bilateral and symmetric distribution of denuded area in an s-shaped broad band with sharply demarcated borders, which is shown in our case mainly at the extremities. The presentation of dystrophic epidermolysis bullosa with congenital localized absence of skin (Bart's Syndrome) is very rare, with only five cases described in the published literature [9]. Other author mentioned three cutaneous manifestation of Bart`s Syndrome: congenital absence of skin, mucocutaneous blistering, and nail abnormalities in six reported cases **[**8]**.** Congenital localized absence of skin has been considered as subtype of aplasia cutis congenita, which is a heterogeneous group of disorders in which localized or wide spread areas of skin are absent at birth. Diagnosis is primarily by clinical features. The causes of this entity vary and include: genetic factors, teratogens, compromised vasculature to the skin and trauma. The condition may be associated with epidermolysis bullosa, specific teratogens or intrauterine infection, or it may occur in the presence of chromosomal abnormalities [10]**.** These possible causes were not excluded completely in our case. Intrauterine infection studies (Toxoplasmosis, Rubella, Cytomegalo virus, Hepatitis, Herpes simplex) were done and was found normal. Bart's Syndrome was thought to be a new variant of Epidermolysis bullosa and Inherited as an Autosomal dominant[11] trait as published in initial series of cases but subsequently it was shown that this is not true and congenital absence of skin can be in association with various forms of EB as well as various other diseases .

CONCLUSIONS

Although the term Bart's Syndrome was originally a useful concept, it was found that physicians may incorrectly assume that Bart's Syndrome and inherited Epidermolysis bullosa are mutually exclusive diseases, where as Bart's Syndrome actually just describes the co–occurrence of congenital localized absence of skin and inherited Epidermolysis

bullosa. It is considered as a subtype of dominantly inherited dystrophic epidermolysis bullosa which needs genetic counseling for the affected families.

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